3-Hydroxy-3-Methylglutaryl -CoA (HMG-CoA) Lyase Deficiency

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of 3-hydroxy-3-methylglutaryl-CoA (HMG-CoA) lyase deficiency. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of HMG-CoA lyase deficiency for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner are carriers for HMG-CoA lyase deficiency, each of your children has a 1 in 4 (25%) chance to have the condition.

3-Hydroxy-3-Methylglutaryl-CoA (HMG-CoA) Lyase Deficiency Explained

What is 3-Hydroxy-3-Methylglutaryl-CoA (HMG-CoA) Lyase Deficiency?

HMG-CoA lyase deficiency is an uncommon inherited disorder in which the body cannot process a particular protein building block called leucine, and the body is prevented from making ketones, which are compounds that are used for energy during periods without food (fasting). The signs and symptoms of HMG-CoA lyase deficiency usually appear within the first year of life and include episodes of vomiting, diarrhea, dehydration, lethargy, and weak muscle tone (hypotonia). During an episode, blood sugar levels can become dangerously low, and a buildup of harmful compounds can cause the blood to become too acidic (metabolic acidosis). If untreated, the disorder can lead to breathing problems, convulsions, coma, and death. Episodes are often triggered by an infection, fasting, strenuous exercise, or other types of stress.

Prognosis

Prognosis is good with early diagnosis and prompt treatment. However, this condition can be fatal in approximately 20 percent of cases. Repeated crises may result in brain damage and significant learning/intellectual disabilities. Symptoms typically become milder after childhood; however, long-term effects may include heart damage, pancreatitis, vision loss, hearing loss, and intellectual disability.

Treatment

Treatment includes avoiding fasting, feeding with a low-leucine diet, medications, and prompt attention during metabolic crises. Individuals are often followed by a physician specializing in metabolism.

Resources

Organic Acidemia Association
https://www.oaanews.org/
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/3-hydroxy-3-methylglutaryl-coa-lyase-deficiency#inheritance
National Society of Genetic Counselors
https://www.nsgc.org/

801 Broadway Ave NW, Suite 203, Grand Rapids, MI 49504 • Phone: (855) 776-9436 • Fax: (616) 710-4667 info@nxgenmdx.com
nxgenmdx.com

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